the orphan disease center

MILLION DOLLAR BIKE RIDE
RAISING MONEY FOR RARE DISEASE RESEARCH

Research Focus Areas:

ZC4H2 Associated Rare Disorders (ZARD) is an ultra-rare genetic condition with central and peripheral nervous system involvement caused by pathogenic variant of the ZC4H2 gene. ZC4H2 is located on the X chromosome and encodes the ZC4H2 (zinc finger C4H2-type containing) protein essential for normal development. ZARD can manifest in a broad range of clinical severity. Clinical presentations of affected individuals who carry the same pathogenic ZC4H2 gene variant can vary within families and between families. Males and females can be affected. To date, approx. 250 cases have been diagnosed worldwide.

Among the many symptoms affecting an individual with ZARD, neuromuscular manifestations (impaired movement, mobility and orthopedic disorders) are consistently ranked within the ZARD community as the most impactful in terms of diminishing quality of life.
There is currently very limited understanding on the role of the ZC4H2 gene and its protein in the development and function of the human muscular system. The focus for this grant opportunity is to understand the impact of pathogenic ZC4H2 variants in the physiology and function of the human skeletal muscles.

For this purpose, **one grant of $61,815**, will be offered to research projects on:

Studies on the pathology, physiology, morphology, anatomy and neural activation of human skeletal muscles affected by pathogenic mutations of the ZC4H2 gene: the studies may involve in-vivo, in-situ and/or in-vitro human materials and in-vivo and in-vitro animal materials. The in-situ and in-vivo human studies should not be invasive.

Applicants are expected to collaborate with other scientists and clinicians currently or previously involved in ZC4H2 research, and should include a statement on resource sharing in their proposal. Applicants are encouraged to use existing tools (e.g., existing viable and validated animal models, antibodies, fibroblasts, LCLs, iPSCs) and to contact the ZC4H2 Research Foundation (info@zc4h2foundation.org) with any questions about these resources.

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